The use of gene tests to detect hereditary predisposition to cancer: economic considerations

Brown M L, Kessler L G

Record Status
This is a critical abstract of an economic evaluation that meets the criteria for inclusion on NHS EED. Each abstract contains a brief summary of the methods, the results and conclusions followed by a detailed critical assessment on the reliability of the study and the conclusions drawn.

Health technology
Genetic testing to detect hereditary predispositions to cancer (colon cancer).

Type of intervention
Screening.

Economic study type
Cost-effectiveness analysis.

Study population
A hypothetical, representative cohort of the general population with an average age of 25 years.

Setting
Community. The economic study was carried out in the USA.

Dates to which data relate
The dates associated with the effectiveness data were not stated. The resource use data were derived partly from studies published in 1991 and 1994. The price year was not clearly stated.

Source of effectiveness data
Evidence for final outcomes were based on opinion.

Modelling
A model was used in estimating both lifetime benefits and costs associated with screening. The model used population-wide data on prevalence and incidence rates of disease for the USA, and included costs and effects associated with screening, further intensive diagnostic follow-up for positive cases, and treatment of disease. Such calculations started from the age 25.

Methods used to derive estimates of effectiveness
Estimates of effectiveness were based on authors’ assumptions.

Estimates of effectiveness and key assumptions
The authors made the following assumptions which underlie their estimate of the effectiveness:

(1) all gene tests are given at 25 years of age;
(2) the lifetime risk of colorectal cancer in general population is 6%;

(3) the median survival from colorectal cancer is 8 years;

(4) penetrance of the HNPCC genotype is 80%;

(5) the median age of diagnosis of HNPCC-related colorectal cancer is 45 years of age;

(6) the average number of years of life gained through the intervention is 20 years (9.76 years when discounted at 5%); and,

(7) implicit in the base case calculations, a 100% rate of accuracy of genetic testing.

**Measure of benefits used in the economic analysis**

Life-years saved (discounted) was the outcome measure used in the economic analysis, the estimate being derived from a model.

**Direct costs**

Whilst costs were discounted, the quantities of resource use were not reported separately from the costs. Costs considered in the analysis were as follows: the unit cost of genetic testing for HNPCC, including counselling, and the diagnostic-treatment regimen (periodic colonoscopy and colonoscopic removal of detected adenomas) that would follow a positive test and treatment of disease given no intervention. The source of some of these costs was information from previously published studies from 1991 and 1994. Total costs were estimated using a model. On the other hand, the costs associated with colonoscopy-induced perforation of the large bowel were not considered. The price year was not clearly stated.

**Currency**

US dollars ($).

**Sensitivity analysis**

One way and multi-way sensitivity analyses were carried out on both the effectiveness and cost data. The main parameters investigated were the prevalence rate, the cost of genetic testing, the life expectancy of the average person treated for HNPCC, the penetrance of the HNPCC genotype, and the discount rate.

**Estimated benefits used in the economic analysis**

The estimated benefits were not explicitly presented in the paper. However, following the formulae provided by the authors, one can calculate the implicit estimates in the results actually reported. The undiscounted life years gained per patient screened ranged from 0.00608 to 0.184. The corresponding figure for discounted life-years gained with screening relative to no screening ranged from 0.00297 to 0.0898. The side-effects of treatment were not considered in this analysis.

**Cost results**

Although not stated, one can follow the authors in calculating the total cost estimates per patient screened, as above. The expected discounted cost per patient screened ranged from $989.4 to $1,041.9 depending on the prevalence of HNPCC gene in the population.

**Synthesis of costs and benefits**

The costs per life-year saved (with costs and benefits both discounted at 5%) ranged from $333,000 to $11,000.
depending on the value of prevalence of HNPCC used, which in turn ranged from 0.00038 to 0.0115. Unless the prevalence of the genotype and classic syndrome is in the range of 50 cases per 10,000 or more people, very favourable assumptions (specifically about a low cost of screening and a high efficacy of intervention) must be made for population-wide genetic testing to be cost-effective (below $50,000). The price year for the figures above was not clearly stated.

Authors’ conclusions
This preliminary analysis suggests that population-wide genetic testing for HNPCC can be cost-effective only if a fairly restrictive set of assumptions are met. The analysis highlights the urgent need for carefully controlled studies to determine the prevalence and the degree of heterogeneity of HNPCC in a large representative population.

CRD Commentary
As the authors state, their results are simplified and are intended for illustrative purposes only. Despite its consequently limited scope, the study provides some helpful methodological pointers for further research in the area of genetic testing.

Implications of the study
The authors pointed to areas where further research is required in order to improve on the quality of the available cost-effectiveness data. These areas are as follows: studies of the prevalence of high-risk cancer syndromes and associated gene mutations in the population; studies of the accuracy and costs of identifying high-risk families by methods other than genetic testing; and costing studies of genetic testing as a function of scale of testing and the number and distribution of mutations.

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